CURRICULUM VITAE

Wendy Jewell Introne, M.D.

Current Position

Staff Clinician
Office of the Clinical Director
National Human Genome Research Institute
National Institutes of Health
10 Center Dr., Building 10 CRC, Room 3-2541
Bethesda, MD 20892
November 2004 - Present

Previous Position

Senior Instructor
Division of Pediatric Genetics
Strong Memorial Hospital
601 Elmwood Ave. Box 777
Rochester, NY 14642
May 2003 – July 2004

Education

University of New Mexico
Albuquerque, New Mexico
B.S. in Biology, May, 1991

University of New Mexico School of Medicine
Albuquerque, New Mexico
M.D., May, 1995

Graduate Medical Education

Pediatric Intern, June 1995 – June 1996
Children's National Medical Center
Washington, D.C.
Pediatric Resident, July 1996 – June 1998
Children's National Medical Center
Washington, D.C.

Medical and Clinical Biochemical Genetics Fellow, July 1998 – June 2001
Medical Genetics Branch, National Human Genome Research Institute and
Heritable Disorders Branch, National Institute of Child Health and Human Development
National Institutes of Health, Bethesda, MD

Certification and Licensure

American Board of Medical Genetics, Clinical Genetics, 2002 - Present
American Board of Medical Genetics, Clinical Biochemical Genetics, 2002 - Present
American Board of Pediatrics, 1998 - Present

District of Columbia Medical License, 1996 - Present
New York State Medical License, 2003
Pediatric Advanced Life Support

Honors and Awards

NHGRI Merit Award, 2008
Alpha Omega Alpha, 1994
P.E.O. Scholar Award, 1994
McGraw-Hill Book Award, 1992
Excellence in Morphology Award, 1992
Phi Beta Kappa, 1991
Phi Kappa Phi Honor Society, 1991

Membership in Professional Societies

Society of Inherited Metabolic Disorders, 2003 - Present
American Academy of Pediatrics, 1995 – Present
Member, Section on Genetics and Birth Defects

Clinical Research Experience

2007 – Current, Associate investigator on National Institutes of Health clinical protocol number 07-HG-0076 – *A Phase One Treatment Trial of the Circadian Sleep Disturbance in Smith-Magenis Syndrome (SMS)*

2005 – Current, Associate investigator on National Institutes of Health clinical protocol number 05-HG-0076 – *Long-Term Clinical Trial of Nitisinone in Alkaptonuria*

2005 – 2008, Associate investigator on National Institutes of Health clinical protocol number 05-HG-0004 – *Clinical Investigations into Hutchinson-Gilford Progeria Syndrome*

2004 – Current, Associate investigator on National Institutes of Health clinical protocol number 01-HG-0109 – *Natural History Study of the Clinical & Molecular Manifestations of Smith-Magenis Syndrome (SMS)*

2000 - 2001, 2004 – Current, Associate investigator on National Institutes of Health clinical protocol number 00-CH-141 –*Clinical, Biochemical and Molecular Investigations into Alkaptonuria*

2000-2001, 2004 – Current, Associate investigator on National Institutes of Health clinical protocol number 00-CH-153 – *Investigations into Chediak-Higashi Syndrome and Related Disorders*

Invited Lectures

Alkaptonuria Society Medical Research Conference
"*Nitisinone Treatment in Alkaptonuria: From darkness into light*"
Liverpool, UK, November, 2008

Alkaptonuria Society Patient Meeting
"Nitisinone Treatment in Alkaptonuria: From darkness into light"
Liverpool, UK, November, 2008

Baltimore Genetics Group
"Putting the Brakes on Accelerated Aging: Hutchinson-Gilford Progeria syndrome"
Baltimore, MD, May, 2008

National Institutes of Health Clinical Center Grand Rounds
"Putting the Brakes on Accelerated Aging: Hutchinson-Gilford Progeria syndrome"
Bethesda, MD, February, 2008

Xeroderma Pigmentosum and Other Diseases of Human Premature Aging and DNA Repair: Molecules to Patients
"Characterizing the Phenotype of Hutchinson-Gilford Progeria Syndrome (HGPS)"
Landsdowne, VA, September, 2006

PRISMS
"SMS across the Lifespan"
Cincinnati, OH, April, 2005

Department of Pediatrics Continuing Professional Education Conference
University of Rochester School of Medicine and Dentistry
"Inborn Errors of Metabolism: A practical approach."
Rochester, NY, April, 2004

Department of Pediatrics Grand Rounds
University of Rochester School of Medicine and Dentistry
"MCAD: Lessons learned from a year of newborn screening."
Rochester, NY, January, 2004

Department of Pediatrics Grand Rounds
Rochester General Hospital
"MCAD: Lessons learned from a year of newborn screening."
Rochester, NY, January, 2004

Rehabilitation Medicine Grand Rounds
National Institutes of Health
"Clinical, Molecular, and Biochemical Aspects of Alkaptonuria"
Bethesda, MD, February, 2001

Rheumatology Grand Rounds
National Institutes of Health
"Clinical, Molecular, and Biochemical Aspects of Alkaptonuria"
Bethesda, MD, January, 2001

University of New Mexico Medical Student Research Day
"CRP-Mediated Synthesis of IL-1 and TNF-alpha by Monocytic Cell Lines and the Role of Second Messengers"
Albuquerque, NM, January, 1993

Meeting Presentations/Abstracts

Clinical and Cellular Correlations in Chediak-Higashi Syndrome. W Westbroek, **WJ Introne**, I Manoli, GA Golas, T Vilboux, DA Adams, D Maynard, M Huizing, WA Gahl. Poster presentation. American Society of Human Genetics, November 2008.

Idiopathic Nephrocalcinosis: Possible Genetic Causes. G Nesterova, **WJ Introne**, GA Golas, C Cicone, M Huizing, WA Gahl. Poster presentation. American Society of Human Genetics, November, 2008.

Correlations of Impairments and Functional Limitations in Alkaptonuria. MB Perry, **WJ Introne**, GP Furst, WA Gahl, LH Gerber. Poster presentation. American Academy of Physical Medicine and Rehabilitation, November 2008.

Felix the Double Helix: Fostering an Interest in Science by Teaching Elementary School Students about DNA. MA Merideth, AM Garcia, TC Markello, HM Dorward, DM Krasnewich, WA Gahl, **WJ Introne**. Poster presentation. American College of Medical Genetics, March 2008.

Tolerance of Elevated Tyrosine Levels in Patients with Alkaptonuria Receiving Nitisinone. **WJ Introne**, MA Kayser, ET Tsilou, KE O"Brien, J Bryant, I Bernardini, WA Gahl. Poster presentation. Society of Inherited Metabolic Disorders, March 2008.

Hutchinson-Gilford Progeria Syndrome (HGPS): Comprehensive characterization of 15 children. MA Merideth, **WJ Introne**, LB Gordon, MB Perry, SB Clauss, V Sachdev, CK Zalewski, CC Brewer, J Kim, JC Graf, ACM Smith, LH Gerber, JA Yanovski, DL Domingo, TC Hart, FS Collins, EG Nabel, RO Cannon, WA Gahl. Platform presentation, American Society of Human Genetics, October, 2007.

Fractional Excretion of Homogentisic Acid is Reduced in Alkaptonuria Patients Treated with Nitisinone (Orfadin). Kayser MA, **Introne WJ**, O'Brien K, Bernardini I, Kleta R, Gahl WA. Poster presentation, American Society of Human Genetics, October 2007.

Felix the Double Helix: Teaching Elementary Students about DNA. HD Edwards, **WJ Introne**, AM Garcia, TC Markello, HM Dorward, MA Kayser, DM Krasnewich, WA Gahl, MA Merideth. Poster presentation, American Society of Human Genetics, October, 2007.

Correlations of functional limitations in Hutchinson-Gilford progeria. MB Perry, **WJ Introne**, M Merideth, GP Furst, WA Gahl, LH Gerber. Arch Phys Med Rehab. 2007; 88(11):E46.

Auditory Phenotype of Hutchinson-Gilford Progeria Syndrome. Zalewski C, Merideth M, **Introne W,** Gordon L, Smith ACM, Kim HJ, Gahl W, Brewer C. Poster presentation, American Academy of Audiology, April, 2007.

Hutchinson-Gilford Progeria syndrome: oral-craniofacial phenotypes. Domingo DL, Trujillo MI, Guadagnini JP, Gordon L, Merideth MA, **Introne WJ**, Gahl WA, and Hart TC. Poster presentation, American and Canadian Associations for Dental Research, March, 2007.

Hearing Loss in Children with Hutchinson-Gilford Progeria Syndrome
MA Merideth, **WJ Introne**, C Zalewski, C Brewer, LB Gordon, ACM Smith, HJ Kim, WA Gahl. Poster presentation, American College of Medical Genetics, March, 2007.

Immunologic abnormalities in Smith-Magenis syndrome: What is the role of TNFRSF13B? **Introne WJ**, Jurinka A, Krasnewich D, Candotti F, Edwards H, Ciccone C, Huizing M, Smith ACM. Platform presentation, Smith-Magenis Syndrome Research Roundtable, March, 2007.

Hutchinson-Gilford Progeria Syndrome (HGPS): Clinical Presentation, Diagnosis and Consistency of Phenotype in 15 Children. MA Merideth, **WJ Introne**, LB Gordon, M

Perry, M Turner, S Clauss, V Sachdev, J Graf, ACM Smith, J Reynolds, C Brewer, C Zalewski, J Kim, LH Gerber, JA Yanovski, R Cannon, WA Gahl. Poster presentation, National Human Genome Research Institute, November, 2006.

Impairments and Functional Limitations in Hutchinson-Gilford Progeria. MB Perry, **WJ Introne**, M Merideth, LB Gordon, WA Gahl, LH Gerber. Arch Phys Med Rehab. 2006; 87(11):E23.

Hutchinson-Gilford Progeria Syndrome (HGPS): Consistency of the phenotype in 15 children. **Introne WJ**, Merideth M, Gordon L, Perry M, Turner M, Clauss S, Sachdev V, Graf J, Smith A, Gerber L, Reynolds J, Yanovski J, Cannon R, Gahl, W. Poster presentation, American Society of Human Genetics, October, 2006.

Clinical Presentation and Diagnosis of Patients with Hutchinson-Gilford Progeria Syndrome. Merideth M, **Introne W**, Gordon L, Smith A, Gahl, W. Poster presentation, American Society of Human Genetics, October, 2006.

Is Disease Severity in Alkaptonuria modified by a common SNP in the organic anion transporter MRP4/ABCC4? Kayser MA, Suwannart P, **Introne W**, Austin HA, Tuchman M, Tinloy B, Klein C, O'Brien K, Bernardini I, Gahl WA, Kleta R. Poster presentation, American Society of Human Genetics, October, 2006.

Characterizing the Phenotype of Hutchinson-Gilford Progeria Syndrome (HGPS). **WJ Introne**, MA Merideth, LB Gordon, M Perry, M Turner, S Clauss, V Sachdev, J Graf, ACM Smith, JC Reynolds, C Brewer, C Zalewski, J Kim, JA Yanovski, LH Gerber, R Cannon, WA Gahl. Presentation, Xeroderma Pigmentosum and Other Diseases of Human Premature Aging and DNA Repair: Molecules to Patients, September, 2006.

Hutchinson-Gilford Progeria Syndrome Objective Clinical Assessment Strategies: How will we know if treatments are improving disease? Gordon LB, **Introne W**, Merideth M, Smith A, Graf J, Reynolds J, Yanovski J, Perry M, Gerber L, Brewer C, Kim H, Zalewski C, Gahl, W. Oral presentation, The Progeria Research Foundation InternationalWorkshop on Progeria, November, 2005.

Immunologic Abnormalities in Smith-Magenis syndrome (del 17p11.2)
Introne W, Jurinka A, Krasnewich D, Candotti F, Smith ACM. Poster presentation, American Society of Human Genetics, October, 2005.

Expanded Newborn Screening for Medium Chain Acyl-CoA Dehydrogenase (MCAD) Deficiency in Western New York. **Introne WJ**, DeLuca J, Arnold GA. Poster presentation, Society of Inherited Metabolic Disorders, March, 2004.

Detection of Hartnup's Disorder in an Alkaptonuria Sibship. Bernardini I, **Introne W**, Kleta R, Fitzpatrick DL, Gahl WA. *American Journal of Human Genetics*, 2001; 69:486/1784.

The Ochronosis of Alkaptonuria is Accelerated by Renal Disease. **Introne WJ**, Bernardini I, Gahl WA. Poster presentation, Society of Inherited Metabolic Disorders, March, 2001.

Homogentisic Acid Interferes with Creatinine Measurement and the Assessment of Renal Function in Alkaptonuria Bernardini I, **Introne WJ**, Gahl WA. Poster presentation, Society of Inherited Metabolic Disorders, March, 2001.

Alkaptonuria: New studies of an old disease. **Introne WJ**, Rausche M, Anikster Y, Gilbert F, Gahl WA. *American Journal of Human Genetics*, 2000; 67:289/1590.

Clinical manifestations in three American adult patients with Carbohydrate-deficient Glycoprotein Syndrome. Krasnewich D, Orvisky E, Goker O, **Introne W**, Peters K, Dietrich K, Ginns E, Sidransky E. *American Journal of Human Genetics*, 1999; 65:A424/2402.

False Positive Imaging Studies in Neck and Back Pain. **IntroneWJ***,* Bicknell J. Poster presentation, New Mexico Chapter of the American College of Physicians, December, 1994.

Bibliography

Domingo DL, Trujillo MI, Council SE, Merideth MA, Gordon LB, Wu T, **Introne WJ**, Gahl WA, Hart TC. Hutchinson-Gilford progeria syndrome: oral and craniofacial phenotypes. Oral Dis. Apr;15(3):187-95. Epub Feb 19, 2009.

Introne WJ, Westbroek W, Golas GA, Adams D (March 2009) Chediak-Higashi syndrome in: *GeneReviews at GeneTests: Medical Genetics Information Resource [database online].* Copyright, University of Washington, Seattle, 1997-2008. Available at http://www.genetests.org.

MA Merideth, LB Gordon, S Clauss, V Sachdev, ACM Smith, MB Perry, CC Brewer, C Zalewski, HJ Kim, B Solomon, BP Brooks, LH Gerber, ML Turner, DL Domingo, TC Hart, J Graf, JC Reynolds, A Gropman, JA Yanovski, M Gerhard-Herman, FS Collins, EG Nabel, RO Cannon, WA Gahl, **WJ Introne**. Hutchinson-Gilford Progeria Syndrome: Phenotype and Course. *N Engl J Med*, 358(6):592-604, 2008.

WJ Introne, MA Kayser, WA Gahl (updated December 2007) Alkaptonuria in: *GeneReviews at GeneTests: Medical Genetics Information Resource [database online]*. Copyright, University of Washington, Seattle. 1997-2007. Available at http://www.genetests.org.

Kayser MA, **Introne WJ**, Gahl WA. Alkaptonuria in: *The Online Metabolic and Molecular Bases of Inherited Disease*. New York, McGraw-Hill Publishers, Updated November, 2007.

Suwannarat P, Phornphutkul C, **Introne WJ**, Gahl WA (May 2003) Alkaptonuria. In: *GeneReviews at GeneTests: Medical Genetics Information Resource [database online]*. Copyright, University of Washington, Seattle, 1997-2003. Available at http://www.genetests.org.

Phornphutkul C, **Introne WJ,** Perry MB, Bernardini I, Murphey MD, Fitzpatrick DL, Anderson PD, Huizing M, Anikster Y, Gerber LH, Gahl WA. Natural History of Alkaptonuria. *N Engl J Med*. 347(26):2111-2121, 2002.

Introne WJ, Phornphutkul C, Bernardini I, McLaughlin K, Fitzpatrick D, and Gahl WA. Exacerbation of the ochronosis of alkaptonuria due to renal insufficiency and improvement after renal transplantation. *Molecular Genetics and Metabolism*. 77:136-142, 2002.

Introne WJ, and Gahl WA. Alkaptonuria. *NORD Guide to Rare Diseases*. Lipincott publishers, 2001.

Introne W, Boissy RE, Gahl WA. Clinical, molecular, and cell biological aspects of Chediak-Higashi syndrome. *Molecular Genetics and Metabolism*. 68:283-303, 1999.

Jewell WS, Marnell LL, Rokeach LA, and DuClos TW. C-reactive protein (CRP) binding to the sm-D protein of snRNPs. Identification of a short polypeptide binding region. *Molecular Immunology.* 30(8):701-708, 1993.